Anne Claude Tabet

List of Publications by Year
in descending order

22	995	623734 14	642732 23
papers	citations	h-index	g-index

23	23	23	2302
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: A Gradient of Severity in Cognitive Impairments. PLoS Genetics, 2014, 10, e1004580.	3.5	501
2	Duplication of the 15q11-q13 region: Clinical and genetic study of 30 new cases. European Journal of Medical Genetics, 2014, 57, 5-14.	1.3	68
3	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
4	Operative list of genes associated with autism and neurodevelopmental disorders based on database review. Molecular and Cellular Neurosciences, 2021, 113, 103623.	2.2	51
5	Autism multiplex family with 16p11.2p12.2 microduplication syndrome in monozygotic twins and distal 16p11.2 deletion in their brother. European Journal of Human Genetics, 2012, 20, 540-546.	2.8	38
6	Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. Molecular Autism, 2015, 6, 19.	4.9	29
7	De novo deletion of TBL1XR1 in a child with nonâ€specific developmental delay supports its implication in intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2335-2337.	1.2	24
8	Dysregulation of the NRG1/ERBB pathway causes a developmental disorder with gastrointestinal dysmotility in humans. Journal of Clinical Investigation, 2021, 131, .	8.2	24
9	NR4A2 haploinsufficiency is associated with intellectual disability and autism spectrum disorder. Clinical Genetics, 2018, 94, 264-268.	2.0	22
10	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
11	The phenotypic and genetic spectrum of patients with heterozygous mutations in cyclin M2 (CNNM2). Human Mutation, 2021, 42, 473-486.	2.5	21
12	Clinical and molecular delineation of Tetrasomy 9p syndrome: Report of 12 new cases and literature review. American Journal of Medical Genetics, Part A, 2015, 167, 1252-1261.	1.2	20
13	Molecular and clinical delineation of 2p15p16.1 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2081-2087.	1.2	18
14	EFNB2 haploinsufficiency causes a syndromic neurodevelopmental disorder. Clinical Genetics, 2018, 93, 1141-1147.	2.0	18
15	Telomere and Centromere Staining Followed by M-FISH Improves Diagnosis of Chromosomal Instability and Its Clinical Utility. Genes, 2020, 11, 475.	2.4	17
16	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
17	Molecular characterization of a de novo 6q24.2q25.3 duplication interrupting UTRN in a patient with arthrogryposis. American Journal of Medical Genetics, Part A, 2010, 152A, 1781-1788.	1.2	13
18	LEF1 haploinsufficiency causes ectodermal dysplasia. Clinical Genetics, 2020, 97, 595-600.	2.0	11

#	Article	IF	Citations
19	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	3.8	9
20	INTUâ€related oralâ€facialâ€digital syndrome type VI: A confirmatory report. Clinical Genetics, 2018, 93, 1205-1209.	2.0	7
21	Rare and de novo duplications containing <scp>TCF20</scp> are associated with a neurodevelopmental disorder. Clinical Genetics, 2022, 101, 364-370.	2.0	7
22	<scp>EPHA7</scp> haploinsufficiency is associated with a neurodevelopmental disorder. Clinical Genetics, 2021, 100, 396-404.	2.0	3